juvenile hyaline fibromatosis

Juvenile hyaline fibromatosis is a disorder that affects the skin, joints, and bones. Individuals with this condition typically begin to develop signs and symptoms within the first few years of life. Juvenile hyaline fibromatosis is characterized by skin bumps that frequently appear on the hands, neck, scalp, ears, and nose. These skin bumps can also develop in joint creases and the genital region. They vary in size and are sometimes painful. Affected individuals usually develop more skin bumps over time.

Juvenile hyaline fibromatosis is also characterized by overgrowth of the gums (gingival hypertrophy) and joint deformities (contractures) that can impair movement. In addition, affected individuals may grow slowly and have bone abnormalities. People with juvenile hyaline fibromatosis typically have severe physical limitations, but most individuals have normal intelligence and live into adulthood.

Frequency

The prevalence of juvenile hyaline fibromatosis is unknown. About 70 people with this disorder have been reported.

Genetic Changes

Mutations in the *ANTXR2* gene (also known as the *CMG2* gene) cause juvenile hyaline fibromatosis. The *ANTXR2* gene provides instructions for making a protein involved in the formation of tiny blood vessels (capillaries). Researchers believe that the ANTXR2 protein is also important for maintaining the structure of basement membranes, which are thin, sheet-like structures that separate and support cells in many tissues.

The signs and symptoms of juvenile hyaline fibromatosis are caused by the accumulation of a clear (hyaline) substance in different parts of the body. The nature of this substance is not well known, but it is likely made up of protein and sugar molecules. Researchers suspect that mutations in the *ANTXR2* gene disrupt the formation of basement membranes, allowing the hyaline substance to leak through and build up in various body tissues.

Inheritance Pattern

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

Other Names for This Condition

- inherited systemic hyalinosis
- juvenile hyalinosis
- molluscum fibrosum
- Murray syndrome
- Puretic syndrome
- Systemic hyalinosis

Diagnosis & Management

Genetic Testing

- Genetic Testing Registry: Hyaline fibromatosis syndrome
  https://www.ncbi.nlm.nih.gov/gtr/conditions/C2745948/

Other Diagnosis and Management Resources

- GeneReview: Hyalinosis, Inherited Systemic
  https://www.ncbi.nlm.nih.gov/books/NBK1525

General Information from MedlinePlus

- Diagnostic Tests
  https://medlineplus.gov/diagnostictests.html
- Drug Therapy
  https://medlineplus.gov/drugtherapy.html
- Genetic Counseling
  https://medlineplus.gov/geneticcounseling.html
- Palliative Care
  https://medlineplus.gov/palliativecare.html
- Surgery and Rehabilitation
  https://medlineplus.gov/surgeryandrehabilitation.html

Additional Information & Resources

MedlinePlus

- Health Topic: Gum Disease
  https://medlineplus.gov/gumdisease.html
- Health Topic: Skin Conditions
  https://medlineplus.gov/skinconditions.html

Genetic and Rare Diseases Information Center

- Hyaline fibromatosis syndrome
  https://rarediseases.info.nih.gov/diseases/6807/hyaline-fibromatosis-syndrome

Educational Resources

- Orphanet: Juvenile hyaline fibromatosis
  http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=2028

Patient Support and Advocacy Resources

- Resource list from the University of Kansas Medical Center
  http://www.kumc.edu/gec/support/derm.html

GeneReviews

- Hyalinosis, Inherited Systemic
  https://www.ncbi.nlm.nih.gov/books/NBK1525

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28juvenile+hyaline+fibromatosis%5BTIAB%5D%29+OR+%28molluscum+fibrosum%5BTIAB%5D%29+OR+%28puretic+syndrome%5BTIAB%5D%29+OR+%28systemic+hyalinosis%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1080+days%22%5Bdp%5D

OMIM

- HYALINE FIBROMATOSIS SYNDROME
  http://omim.org/entry/228600

Sources for This Summary

- Antaya RJ, Cajaiba MM, Madri J, Lopez MA, Ramirez MC, Martignetti JA, Reyes-Múgica M. Juvenile hyaline fibromatosis and infantile systemic hyalinosis overlap associated with a novel mutation in capillary morphogenesis protein-2 gene. Am J Dermatopathol. 2007 Feb;29(1):99-103.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/17284973
- Dowling O, Difeo A, Ramirez MC, Tukel T, Narla G, Bonafe L, Kayserili H, Yuksel-Apak M, Paller AS, Norton K, Teebi AS, Grum-Tokars V, Martin GS, Davis GE, Glucksman MJ, Martignetti JA. Mutations in capillary morphogenesis gene-2 result in the allelic disorders juvenile hyaline fibromatosis and infantile systemic hyalinosis. Am J Hum Genet. 2003 Oct;73(4):957-66. Epub 2003 Sep 12.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/12973667
  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1180616/
- GeneReview: Hyalinosis, Inherited Systemic
  https://www.ncbi.nlm.nih.gov/books/NBK1525

- OMIM: HYALINE FIBROMATOSIS SYNDROME
  http://omim.org/entry/228600
- Hanks S, Adams S, Douglas J, Arbour L, Atherton DJ, Balci S, Bode H, Campbell ME, Feingold M, Keser G, Kleijer W, Mancini G, McGrath JA, Muntoni F, Nanda A, Teare MD, Warman M, Pope FM, Superti-Furga A, Futreal PA, Rahman N. Mutations in the gene encoding capillary morphogenesis protein 2 cause juvenile hyaline fibromatosis and infantile systemic hyalinosis. Am J Hum Genet. 2003 Oct;73(4):791-800. Epub 2003 Aug 21.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/14508707
  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1180602/
- Rahman N, Dunstan M, Teare MD, Hanks S, Edkins SJ, Hughes J, Bignell GR, Mancini G, Kleijer W, Campbell M, Keser G, Black C, Williams N, Arbour L, Warman M, Superti-Furga A, Futreal PA, Pope FM. The gene for juvenile hyaline fibromatosis maps to chromosome 4q21. Am J Hum Genet. 2002 Oct;71(4):975-80. Epub 2002 Sep 4.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/12214284
  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC378553/

Reprinted from Genetics Home Reference:
https://ghr.nlm.nih.gov/condition/juvenile-hyaline-fibromatosis

Reviewed: December 2008
Published: March 21, 2017

Lister Hill National Center for Biomedical Communications
U.S. National Library of Medicine
National Institutes of Health
Department of Health & Human Services